

A CASE OF AMAUROTIC FAMILY IDIOCY.

By SYDNEY KUH, M.D., Chicago.

The case which I am about to report differs in some essential points from most of the other cases of amaurotic family idiocy heretofore recorded, so that I should have had some hesitation about making the diagnosis had it not been for the pathognomonic changes in the fundus oculi.

The patient, J. E., 13 months old, was brought to my office for examination on the 22nd of October, 1898. Her parents are Jewish, the father is a clerk. The family history is absolutely negative. There is no consanguinity, no alcoholism, no history of nervous or mental disease in the ancestry. The father denies having ever had any symptoms of syphilis. The little girl was the only child, born at full term, and the labor was neither protracted nor unusually difficult, no instruments having been used. During pregnancy the mother's health was good; she suffered no trauma.

The little girl seemed to develop normally until she reached the age of five months; she was breast-fed. At the time just mentioned the mother first noticed that the child could no longer hold up her head. At about the same time the patient's vision began to suffer, though she still seems to notice light. She moves the hands and legs but has very little strength in either, cannot sit up without support, never learned to stand or walk, has made no attempt to talk, does not seem to know her mother any more or to see anything. When spoken to the child will turn towards the source of sound. The hearing appears to be good. At the age of one year our little patient had her first tooth. Obstinate constipation. During the last two months the patient has suffered from spasms which are rapidly increasing in frequency. Since the little girl was six months old she has hardly ever cried.

Status præsens: The child is about normal in size, but very much emaciated. The muscles are all soft and flabby, except in the right arm, in which there is some rigidity; are atrophied all over the body. The facial expression is that of an idiot. The skull is large. The fontanelles are still open. The ears are unusually large, the lobules adherent. The child has a saddle nose. Dr. Haight was kind enough to make a rhinological examination for me, the result of which was negative. The pupils are wide and react to light. Any sudden noise, such as the clapping of hands, slamming of a door, etc., causes generalized spasm (hyperacuity). At times it seemed as though the child noticed a bright light, but we are not cer-

tain about this; objects that were not very bright were surely not seen. The deep reflexes are absent. The internal organs appear normal; there is no swelling of glands; no sign of skin disease.

During the examination the child had a number of spasms, partly due to some noise and then general, partly spontaneous and then limited to certain parts of the body. First the left side of the face began to twitch and both eyes were turned to the left, then there was a series of rhythmical contractions of the extrinsic muscles of the eyes, causing the eyeballs to move from the central line to the left. Shortly after this the convulsions affected the right arm and leg and a little later the left leg became similarly affected.

Ord.: 0.1 each of iodide of potassium and bromide of strontium *t. i. d.*

On November 12 the child was again brought for examination. The mother said that the spasms had become less frequent and less violent and lasted only a shorter time. During the last week there had been hardly any. To-day, however, they are again more frequent. Medicine had not been given regularly. During the last days the child has cried frequently again. Has had seven teeth within the last three weeks. The appetite is good and patient looks as though she had gained some in weight. The pupils are dilated. Pulse-rate 132 a minute. Patient seems brighter. Clapping of the hands, which formerly always produced spasms, has no such effect now; but there are irregular clonic spasms, affecting various parts of the body, during which the eyeballs are generally turned to the left. They seem to arise spontaneously. After that examination the patient was not brought to the office again, but on April 16, 1899, Drs. Haight and Beard and I visited her at her residence. We then found the skull greatly enlarged, broad in the parietal regions and with arched frontal bones. It measured $21\frac{1}{2}$ inches in circumference. The spasms are decidedly less frequent, resemble more the *petit mal* type. The general condition is about the same as at the time of the previous examination; the child now has all of her teeth.

A few days ago I wished to see the little girl again and upon visiting her father heard that she had died three months ago. The paresis had gradually increased until there was total paralysis of all four extremities; the child became more emaciated while the skull increased in size until it measured $22\frac{1}{2}$ inches shortly before the patient's death. The spasms had not become more frequent again.

The points in which this case differs from those reported by

others are, firstly, the great frequency of the epileptic spasms and, secondly, the existence of a pronounced hydrocephalus. While both of these symptoms have been recorded in some other instances, they do not appear to have been as prominent features as they were in my case.

Sachs' theory that amaurotic family idiocy is due to arrested development of nervous tissues seems to me not in accordance with the facts as presented by my case. There appeared to be something more, a distinctly retrogressive and destructive change. A perusal of the observations made by others tends only to strengthen that conviction.

THE APPEARANCES OF THE FUNDUS OCULI.

By CHAS. H. BEARD, M.D.

I must express my gratitude to Drs. Kuh and Patrick for having afforded me the opportunity of studying ophthalmoscopically these two interesting cases and of making a colored drawing in each. The ophthalmoscopic pictures here presented are unique. There is no other in any way similar. Not even that seen in embolism of the central artery of the retina, as has been more than once asserted. When it comes to appearances of the fundus oculi *characteristic* of a general disease or disorder, these stand alone, "grand, gloomy and peculiar." To describe them briefly:

The disc is of normal size and stands out unusually clear-cut as to its borders, and the choroidal ring is specially distinct—*i. e.* free from the obscuring radiate striation normally present in the eyes of children. The lamina cribrosa is slightly veiled, though visible. Contrary to what I had been led to expect, there is not pronounced atrophy of the optic nerve and retina. The outer half of the disc is decidedly blanched, and the retinal vessels are all somewhat reduced in size. The choroid and the hexagonal pigment, wherever visible, appear normal. The great dominating feature of the picture, and that which characterizes this fundus and is absolutely diagnostic of the disease in question, is what one sees in and immediately around the macula lutea. Surrounding the fovea centralis, and concentric with it, though fully two or three times as large, is a

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liver-colored disc. This disc is the center of a zone of grayish-white, which extends for at least two discs diameters horizontally, and somewhat less vertically in every direction from the center, and gradually fades away into the normal red-orange of the eyeground. This livid disc is as clear-cut as a coin—not irregular in outline, as is the case in acute inflammatory conditions where the surrounding retina is infiltrated; nor is it either cherry-red or carmine, as in those other instances, but is distinctly brownish. Instead of coinciding with the fovea in area, as is stated above, it is larger. That is to say, instead of marking the area which is occupied by the cones alone it marks that which is devoid of the ganglion cells.

Another highly distinguishing feature is observed in the character of the whitish zone surrounding the center. This is nebulous rather than cloudy. It is nearly white at the circumference of the liver-colored disc, thence gradually thins away to nothing, but is translucent and shows *some* color throughout. Far from obscuring the retinal vessels which enter it, it only serves to make them more distinct by contrast, so that one is able to trace the tiniest of them right up to the central spot.

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- 46 UN CAS D'AMYOTROPHIE PROGRESSIVE D'ORIGINE TRAUMATIQUE (A Case of Progressive Amyotrophy of Traumatic Origin). F. Sano (Journal de Neurologie, 1899, No. 23, p. 441).

Sano reports the case of a man who was severely injured in the back and thorax. About two months and a half after the accident the right lower limb began to waste and sensation in the part became diminished. Reaction of degeneration was not obtained. The wasting of the limb may have been due to neuritis, although the absence of pain on pressure over the nerves, and of vasomotor and trophic disturbances of the skin, and of disturbance of sensation limited to the distribution of certain nerves, made the diagnosis of progressive atrophy of spinal origin seem more probable. A few cases of spinal atrophy resulting from trauma have been reported. SPILLER.

- 47 STÖRUNGEN DER VASOMOTORENTHÄTIGKEIT UND DER SENSIBILITÄT NACH PERIPHERER TRAUMATISCHER FACIALISLÄHMUNG (Disturbance of the Vasomotor Functions and of Sensation Following Peripheral Traumatic Facial Paralysis). Carl Biehl (Wiener klin. Wochenschrift, 13, 1900, p. 131).

Biehl reports a case in which the left facial nerve was injured by a stab-wound in front of the left ear. The sensation in the distribution of this nerve was much impaired, and perspiration over the left cheek and reddening of this cheek occurred when the patient ate. As the fifth nerve was not injured, Biehl thinks the case proves that the facial nerve contains vasomotor and sensory fibers. SPILLER.